SLC9A6 gene

solute carrier family 9 member A6

Normal Function

The *SLC9A6* gene provides instructions for making a protein called sodium/hydrogen exchanger 6 (Na+/H+ exchanger 6 or NHE6). Na+/H+ exchangers are found in the membranes that surround cells or compartments within cells. These proteins act as channels that allow positively charged sodium atoms (Na+ ions) into the cell or cellular compartment in exchange for positively charged hydrogen ions (H+, also known as protons), which are removed. The exchange of hydrogen ions helps regulate the relative acidity (pH) of the cell or cellular compartment.

The NHE6 protein is found in the membrane of compartments within the cell known as endosomes, which recycle proteins and other cellular materials. The NHE6 protein controls the pH inside endosomes, which is important for the recycling function of these compartments. The NHE6 protein may have additional functions, such as helping to move proteins to the correct location in the cell (protein trafficking).

Health Conditions Related to Genetic Changes

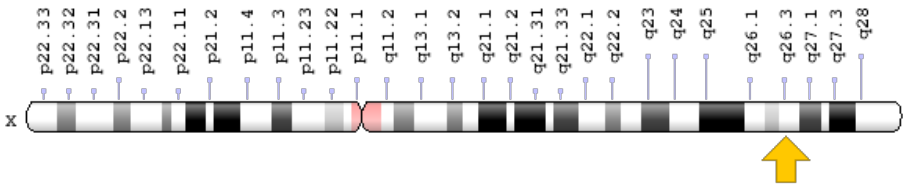
Christianson syndrome

Mutations in the *SLC9A6* gene lead to Christianson syndrome. This condition is characterized by neurological problems, including intellectual disabilities, seizures, and an inability to walk or speak. Mutations in the *SLC9A6* gene typically lead to an abnormally short NHE6 protein that is nonfunctional or that is broken down quickly in cells, resulting in the absence of functional NHE6 channels. As a result, the pH in endosomes is not properly maintained. It is unclear how unregulated endosomal pH leads to neurological problems in people with Christianson syndrome. Some studies have shown that protein trafficking by endosomes is important for learning and memory, but the role of endosomal pH or the NHE6 protein in this process has not been identified.

Chromosomal Location

Cytogenetic Location: Xq26.3, which is the long (q) arm of the X chromosome at position 26.3

Molecular Location: base pairs 135,974,597 to 136,047,269 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- KIAA0267
- Na(+)/H(+) exchanger 6
- NHE6
- SL9A6_HUMAN
- sodium/hydrogen exchanger 6
- solute carrier family 9 (sodium/hydrogen exchanger), member 6
- solute carrier family 9 member 6
- solute carrier family 9, subfamily A (NHE6, cation proton antiporter 6), member 6

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (Fourth Edition, 2002): Specific Proteins are Removed from Early Endosomes and Returned to the Plasma Membrane https://www.ncbi.nlm.nih.gov/books/NBK26870/#A2396

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC9A6%5BTIAB%5D%29+OR+%28%28KIAA0267%5BTIAB%5D%29+OR+%28NHE6%5BTIAB%5D%29+OR+%28sodium/hydrogen+exchanger+6%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- SOLUTE CARRIER FAMILY 9, MEMBER 6
  http://omim.org/entry/300231

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC9A6%5Bgene%5D
- HGNC Gene Family: Solute carriers
  http://www.genenames.org/cgi-bin/genefamilies/set/752
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11079
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/10479
- UniProt
  http://www.uniprot.org/uniprot/Q92581

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